Rapid DNA

DHS Science & Technology Directorate

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Rapid DNA Overview

Reduces Multi-million Dollar Laboratory Processes to One Field Device

- Integration of five forensic lab processes with disposable microfluidic technology.
- Automation allows DHS officers to process samples and receive final results.
- Two U.S. small businesses have commercial devices ready for purchase.

Transport from collection to lab Days/Weeks/Months	Extraction/ Purification 3 hours	Quantification 2 hours	STR Marker Copying 3 hours	Separation and Detection 2 hours	Profile Match Hours/Weeks

10 hours processing reduced to 90 min.

Desktop Rapid-DNA Unit

40% sample cost reduction. System $250K vs. $1M lab.

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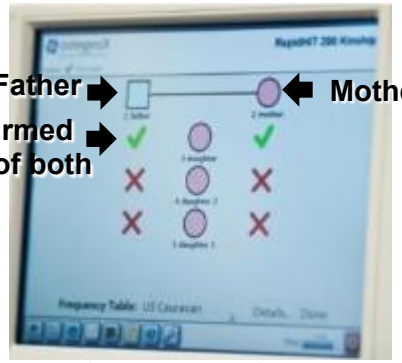
Fully Integrated & Automated

Enables the Decision Maker:

- A new and better capability – Only biometric that verifies family relationships at greater than a 99.5% probability.
- Unique capability – Provides ability in the field to check against criminal and known or suspected terrorist (KST) DNA databases.
- Analyzes trace evidence:
  (i.e., degraded bodies, bones, IED twist ties, tooth brush).
- Ready for field and officer use: ruggedized with an easy interface.
- Compatible with:
  - FBI, Interpol, ANSI/NIST and AABB data formats & reporting standards.

Provides Screening at Speed:

- Microfluidics speeds processes and reduces costs.
- 'Privacy by Design' protection built into the system.
- No special training – fully automated DNA/kinship analysis.
- Disposable sample kit avoids run to run contamination.



Results screen showing family relationships

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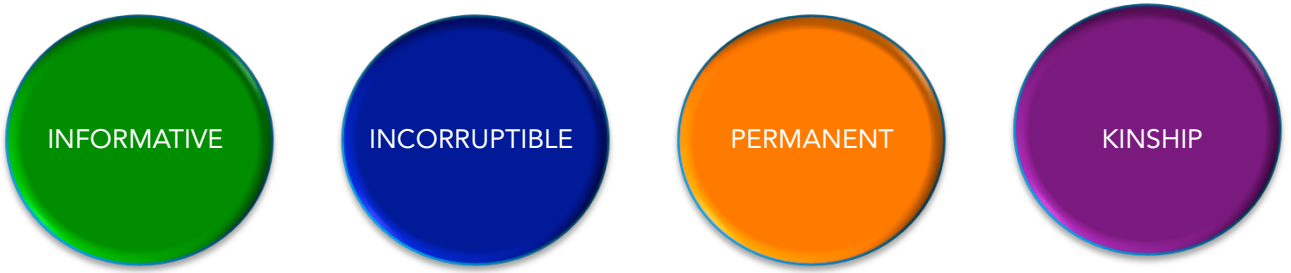
DNA Is The Most Powerful Biometric

More Effective, Informative, And Accurate

- Based on Objective Scientific Principles
- Supported by Statistics, Not Proprietary Algorithms
- Does Not Change Over Time
- Not Subject to Age Restrictions and Reliable After Death
- Effective in Trace Amounts
- Small Data Storage Footprint
- The ONLY Biometric to Verify Biological Relationships
- Privacy is protected by choosing DNA locations that do not reveal any physical traits, race, ethnicity, disease susceptibility, medical information or other sensitive information



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Potential DHS Applications

Immigration:

- Enabling officers to rapidly screen applicant relationships:
  - During refugee interviews and against remote family members.
  - For those putting children up for overseas adoptions.
  - Against federal watch lists/criminal databases.

Border and Enforcement:

- Countering human smuggling and trafficking by verifying relationships of suspicious persons and claimed families.
- Supporting investigations by linking objects to known persons.
- Screening arrested/detained persons against DNA watch lists.

Disaster Recovery and Resilience:

- Medical Examiner daily use for morgue identifications reduces body storage costs and ensures medicolegal staff is ready to deploy.
- Helps resilient communities rapidly recover by identifying victims – even before decontamination – and reuniting families.
- FEMA grants can fund system and consumables purchase.

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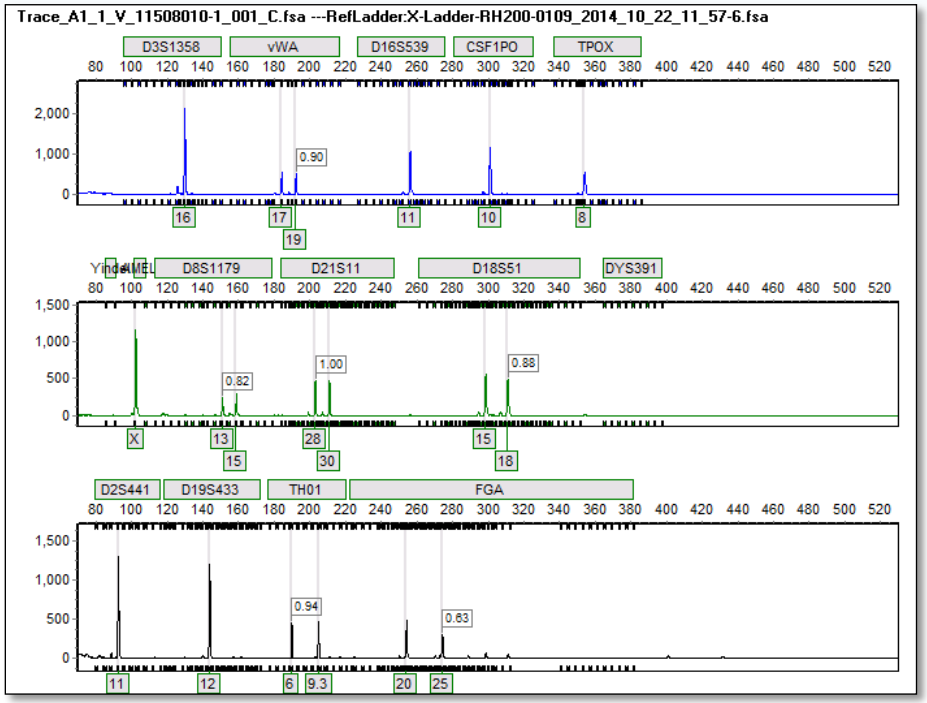
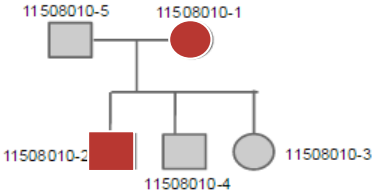
What a DNA Profile Looks Like

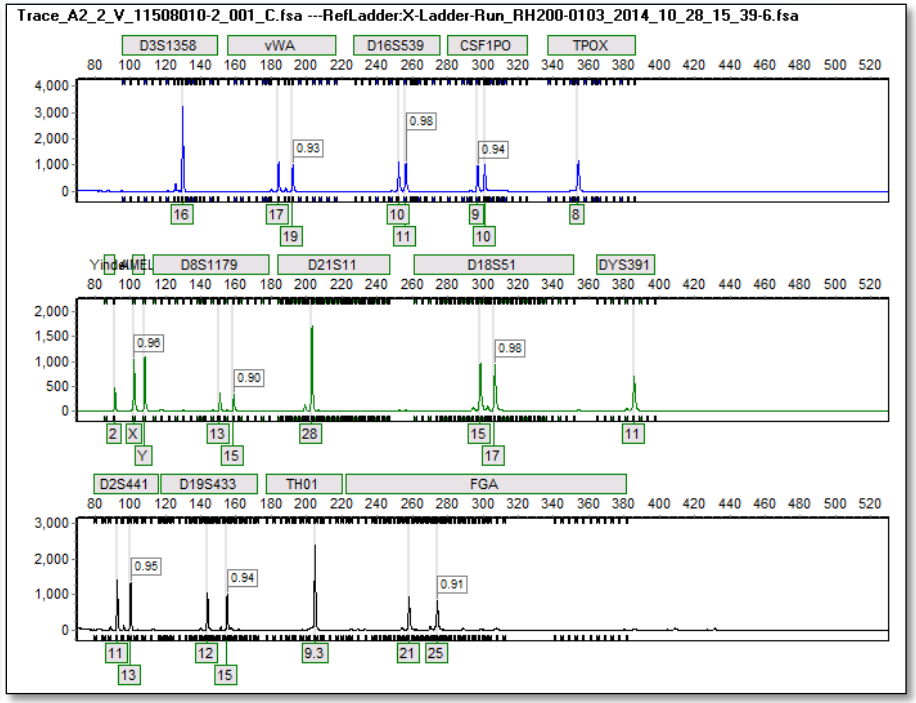
Mother (10-1)



DNA Profile: 16, 16; 17,19; 11, 11; 10,10; 8, 8;
-, -; X, X; 13,15; 28, 30; 15, 18; -, -;
11, 11; 12, 12; 6, 9.3; 20, 25



Child (10-2)



DNA Profile: 16, 16; 17,19; 10, 11; 9,10; 8, 8;
2, 2; X, Y; 13,15; 28, 28; 15, 17; 11, 11;
11, 13; 12, 15; 9.3, 9.3; 21, 25

Note: Only a portion of the profiles are shown

Probability of Maternity = 99.99999996%

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Rapid-DNA Program Metrics

Operational Requirements:

1. Fully automated swab to answer
2. Disposable consumables, pre-loaded with reagents and sealed
3. System cost ≤ $275K, (currently $250K)
4. Cost/sample ≤ $100
   (NetBio currently $270/sample)
   (IntegenX currently $225/sample)
5. Ruggedized for transportation
6. ≤ 6 cu. ft., no side > 30 in., < 110 lbs
   (IntegenX currently 180 lbs.)
7. <1 hr of training for field user
8. Two person setup in ≤ 15 min
9. Reliability of system as specified
   ≥ 1 month routine support interval
10. No routine alignment or calibration
11. 120V, drawing < 5 amps

Sample Analysis:

1. Kinship Verification > 99.5%
2. Answer in <1 hr., Goal = 45 m
   (NetBio 82 min.)
   (IntegenX 115 min.)
3. Simultaneous processing of 5 samples, Goal = 15 samples
   (IntegenX capable of 7)
4. Reagent stable >3 mo. at 20-300C, Goal = 6 mo. at -10-500C
   (NetBio >9 mo. stability)
   (IntegenX = 6 mo. stability but must be refrigerated)
5. DNA extraction & purification comparable to lab methods
6. FBI CODIS compatibility and quality (≥ 16 loci, single base-pair resolution, 500 bp length)
7. DHS Goal: 24 loci for kinship
   (NetBio currently 16 loci)
   (IntegenX currently 24 loci)
8. Processes fresh/dried buccal swabs & other DNA samples prepared manually

Data Analysis:

1. Create & export profiles compatible with CODIS, ANSI/NIST, and expert systems
2. Raw & processed DNA profile data must be provided & stored
3. Bar code reader ~~& GPS receiver that relay position & time to onboard computer~~
4. Generate data file for sample tracking with unique identifier information
5. Software: Windows XP, network connection capability, automated STR allele calling & profile generation, Comms, System control
6. ~~Wireless~~, wired, & USB network connections

Green = Demonstrated
Black = Not yet demonstrated
Red = Below target performance

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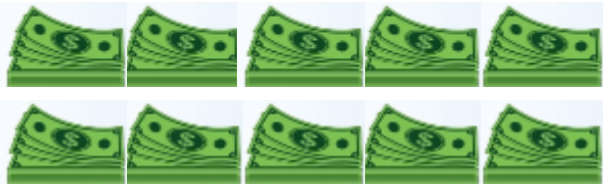
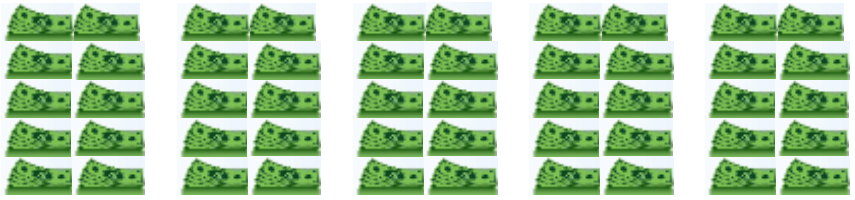
Probability of Relationship Illustration

% Probability of Relationship	Likelihood of Claimed Relationship
50	 Just as likely to be related to each other as to a random person
99	 100 times more likely to be related to each other than to a random person
99.5	 500 times more likely to be related to each other than to a random person

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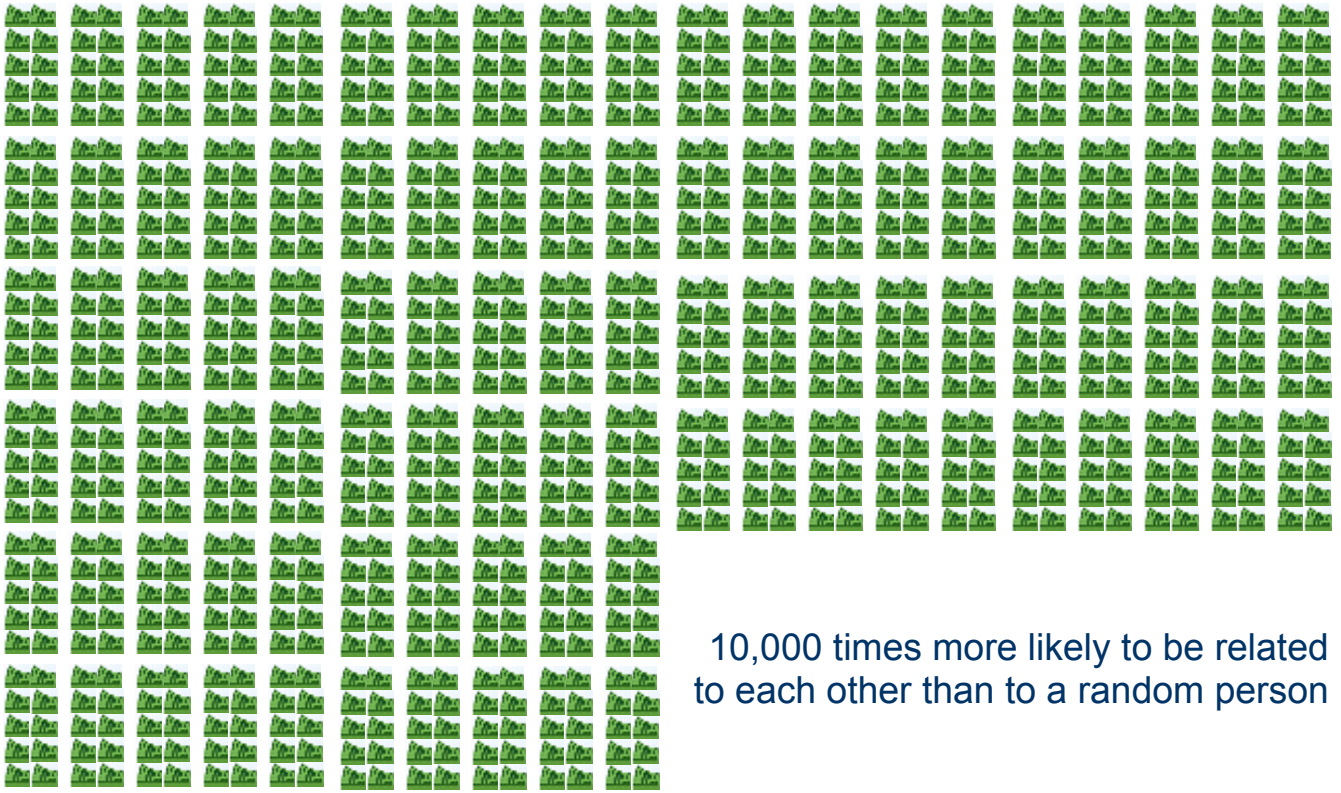
Probability of Relationship

% Probability of Relationship	Likelihood of Claimed Relationship
99.99	

10,000 times more likely to be related to each other than to a random person

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Internal Validation for Kinship

GENERATING PROFILES

- Evaluate that the Rapid DNA instrument produces accurate/reliable allele calls
- Evaluate that the Rapid DNA analysis software correctly identifies profile/data anomalies

EXPORTING PROFILES

- Evaluate the DNA profiles export into the interpretation software correctly

KINSHIP ANALYSIS

- Evaluate the kinship software generates & reports correct statistical calculations

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	Kinship Cases Exchanged with Accredited Laboratory 31 Samples	Allele Call Concordance Study 200 Determinations/ Electropherograms	Artifact/Anomaly Concordance Study 100 Electropherograms	Statistical Concordance Study Spanning DNA Locations/ Alleles/Formulae ~10 Cases
Does Microfluidic Technology Produce Accurate/ Reliable DNA Profiles?	✔			
Does the Expert System Call Alleles Correctly?	✔	✔	✔	
Does the Expert System Accurately Identify Artifacts?		✔	✔	
Are Profiles Exported Properly Into the Kinship Software?	✔	✔	✔	✔
Does the Kinship Software Generate & Report Correct Statistics?	✔			✔

Kinship Buccal Swab Set **DNA Profile Data Test Set** **Kinship Data Test Set**

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Kinship Buccal Validation Set

- 31 Donor Samples collected by Paternity Testing Corporation (PTC) Laboratories (an accredited AABB lab).
- Includes four family groups covering diverse biological associations (e.g. grandparent, aunt/uncle, half-siblings). See next slide.
- Six buccal swabs collected from each person and barcoded with a unique identifier to protect privacy.
- Samples tested at PTC and NIST using conventional DNA methods.
  - DNA data and kinship reports provided to DHS S&T.
- Results presented are from Rapid DNA platform
  - Conventional and Rapid DNA results were concordant.

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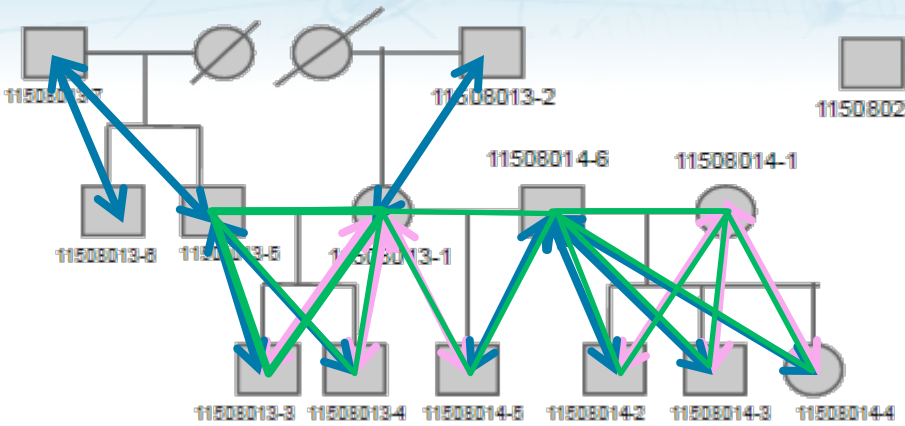
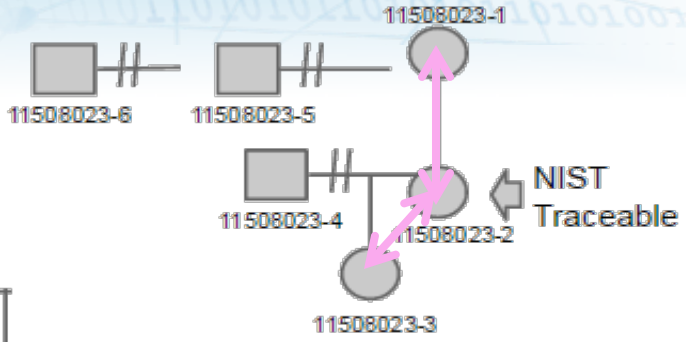
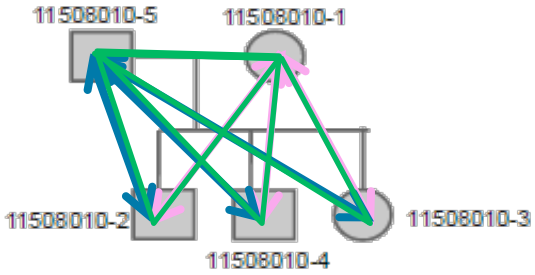
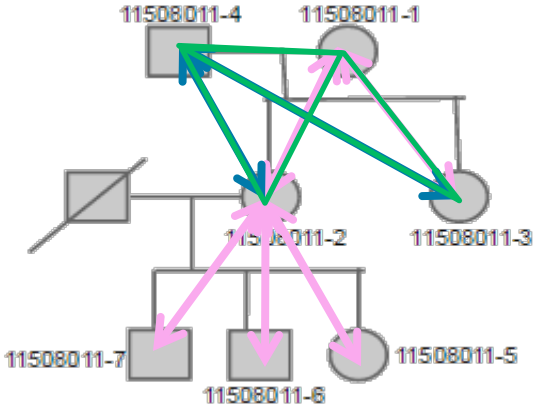
Kinship Validation Samples









↔ = 16 Mother to Child Relationship

↔ = 14 Father to Child Relationship

↔ = 11 Mother/Father/Child Relationships

+12 Full Siblings, 5 Half Siblings,
12 Grandparent/Grandchild, and
5 Aunt/Uncle – Niece/Nephew

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Kinship Associations

Kinship	# Comparisons	# Kinship Evaluations
Mother – Child	16	42
Father – Child	14	
Grandmother – Grandchild	4	
Grandfather – Grandchild	8	
Family Trio (Paternity)	*11*	*33*
Full Siblings	*12*	
Half Siblings	*5*	
Aunt/Uncle – Niece/Nephew	*5*	

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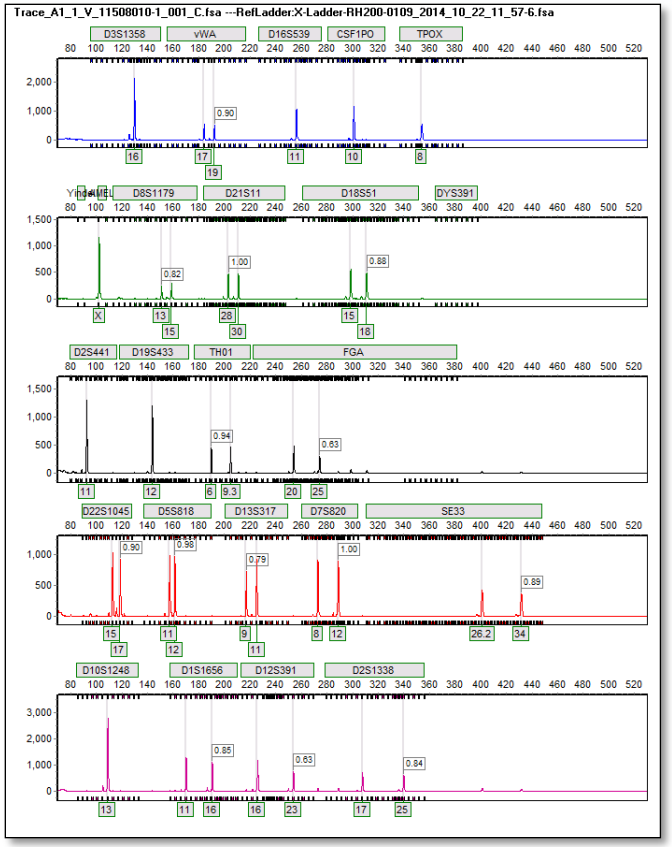
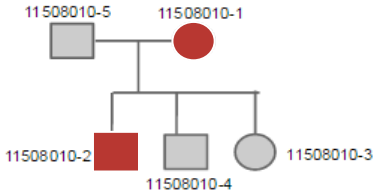
Half of Child's DNA from Each Parent

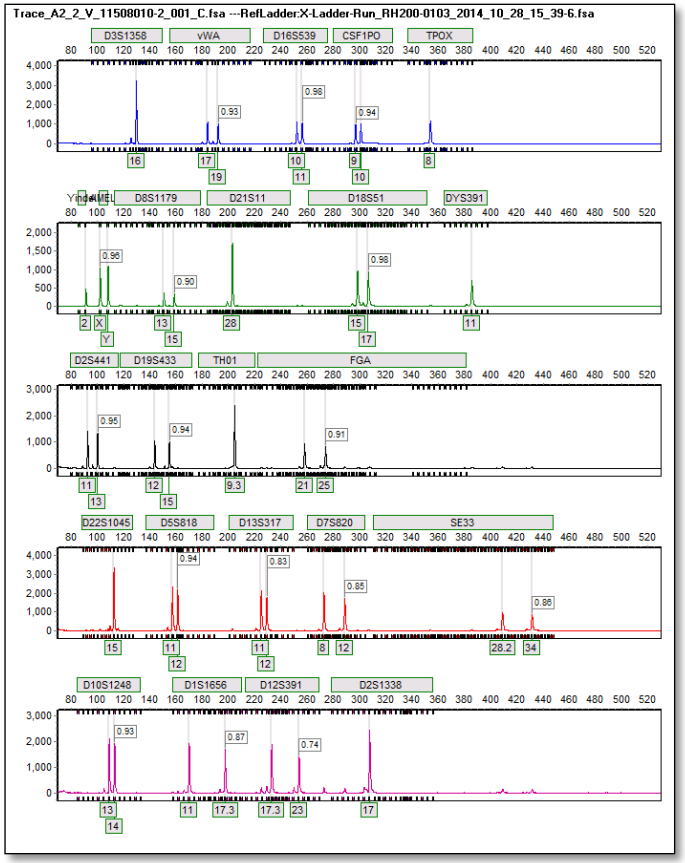
Mother (10-1)





Probability of Maternity
99.99999996%

Child (10-2)



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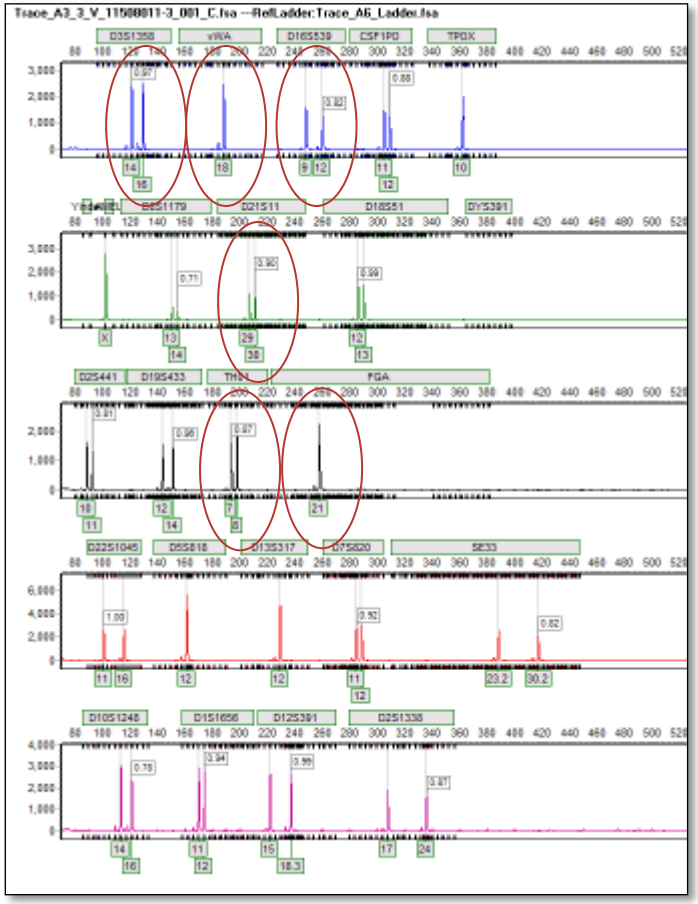
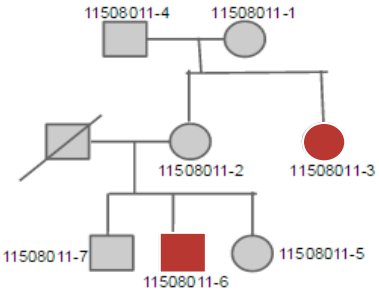
Aunt Can't Falsely Claim to be Mother

Aunt (11-3)

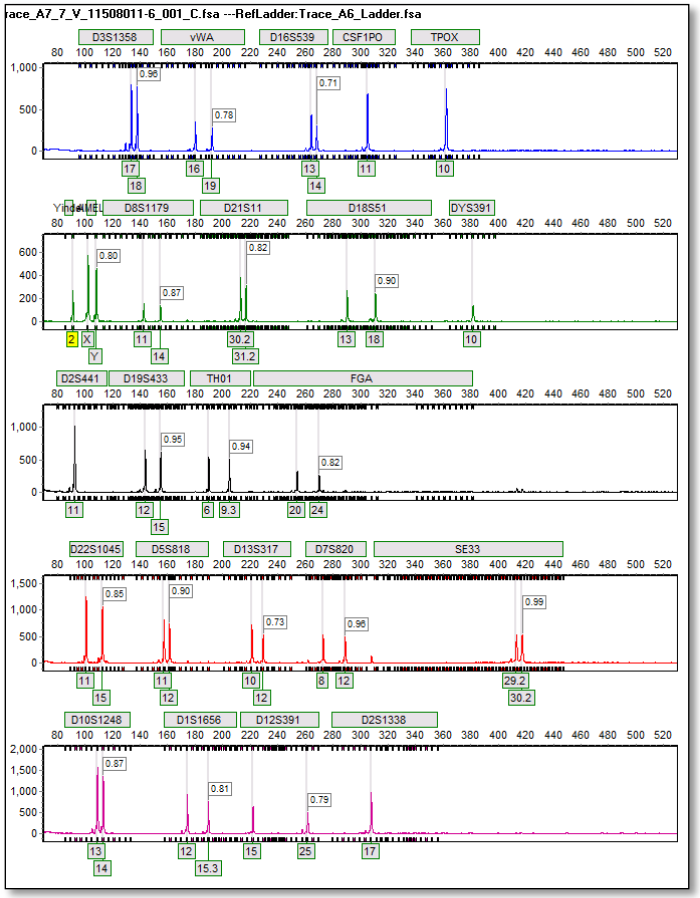




Excluded Match (Six locations don't match):

- D3
- vWA
- D16
- D21
- TH01
- FGA

Child (11-6)



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DHS Kinship - True Claims

PTC Kinship Samples	# of tests	Average Relationship Result	Minimum Result
Mother – Child	16	99.999999997%	99.99995%
Father – Child	14	99.9999998%	99.99998%
Family Trio (Paternity)	11	99.999999998%	99.99998%
Full Siblings	12	99.999999997%	94.84%
Half Siblings	*5*	99.78%	95.53%
Grandmother – Grandchild	4	99.93%*	91.66%
Grandfather – Grandchild	8	98.93%*	8.12%
Aunt/Uncle – Niece/Nephew	*5*	99.21%	30.02%

*When additional family is added for grandparent-grandchild relationships in 8 available tests, the average probability of relationship is 99.99997% with a low of 99.4%

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DHS Kinship - False Claims

PTC Kinship Samples False Claims	# of tests	Average Relationship Result	Maximum Result
Aunt – Child *(Mother Claim)*	3	Exclusion	NA
Uncle – Child *(Father Claim)*	2	Exclusion	NA
Aunt – Uncle – Child *(Mother/Father/Child Claim)*	3	Exclusion	NA
Half Siblings *(Full Siblings Claim)*	5	98.60%*	99.65%
Aunt – Niece (Grandmother – Grandchild Claim)	3	98.34%**	98.48%
Uncle – Nephew (Grandfather – Grandchild Claim)	2	99.52%	98.98%

* False Half Siblings averaged 0.019% when additional siblings are added across 2 tests
** False Grandmother-Grandchild with the mother included averaged 36.44% across 3 tests

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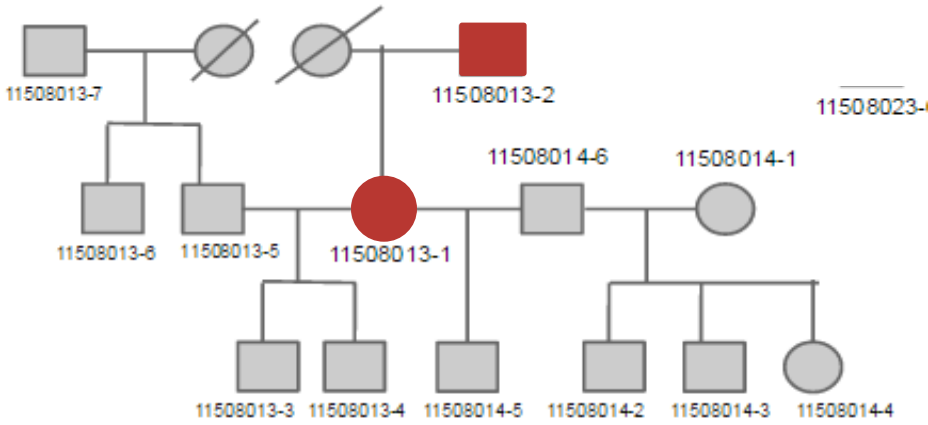
Comparing Population Databases

- Individual 20: 1/2 Asian 1/2 Caucasian (Father)
- Individual 23: 1/4 Asian and 3/4 Caucasian (Daughter)

Population Group	Probability of Relationship
Asian	99.999998%
African American	99.999998%
Caucasian	99.9999996%
Hispanic	99.99999992%



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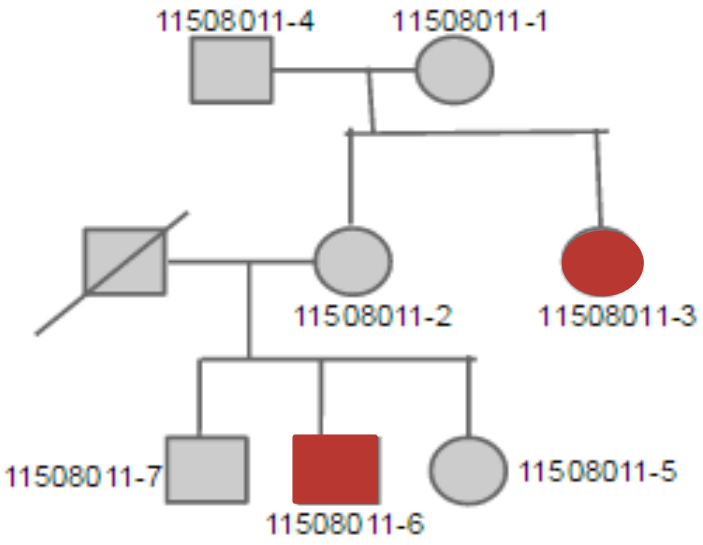
Comparing Population Databases

- Individual 9: Caucasian (Aunt)
- Individual 11: Caucasian (Nephew)

Population Group	Probability of Relationship
Caucasian	98.2%
African American	99.5%
Hispanic	99.3%
Asian	99.92%



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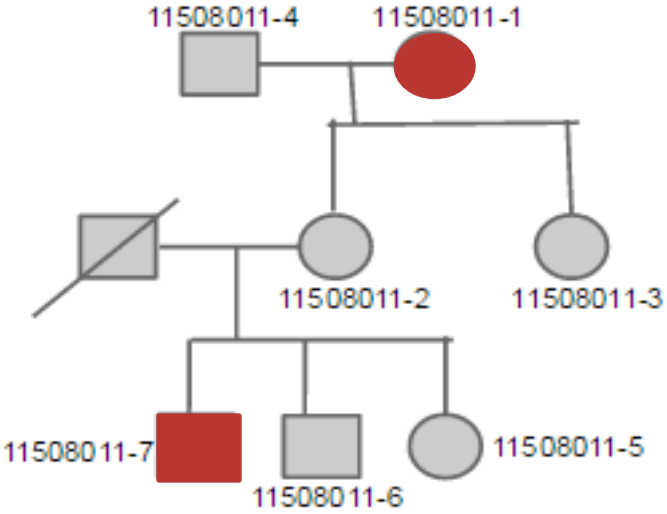
Comparing Population Databases

- Individual 7: Caucasian (Grandmother)
- Individual 10: Caucasian (Grandson)

Population Group	Probability of Relationship
Caucasian	99.87%
Hispanic	99.95%
African American	99.992%
Asian	99.998%



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New Technology is More Robust

Relationship	# of tests	Average Probability of Relationship		
		CODIS 13 Loci	CODIS 20 Loci	24 Loci
Parent-Child	10	99.98%	99.99996%	99.999997%
Full Siblings	5	99.97%	99.99996%	99.999996%
Half Siblings	3	73.64%	73.1%	96.88%
Grandparent-Grandchild	7	74.10%	88.72%	95.88%
Aunt/Uncle-Niece/Nephew	3	23.18%	36.53%	74.82%

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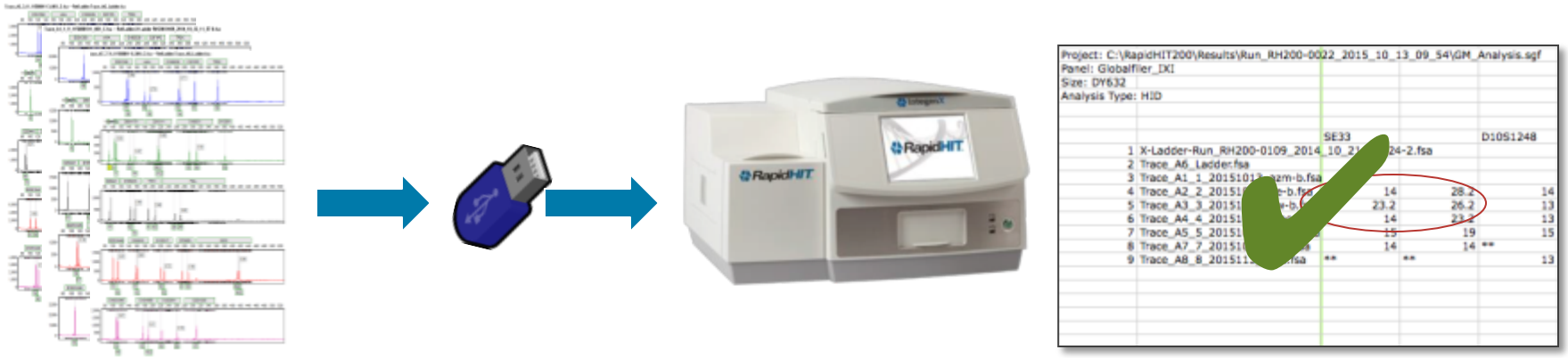
DNA Profile Data Test Set

- S&T seeking to collaborate with Rapid DNA customers to build:
  - 200+ raw .fsa files and associated electropherograms
  - 100 profile anomalies
- Working with Rapid DNA vendors to ingest .fsa files into existing instrument pipeline
- Run every time expert software is updated/modified



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Kinship Data Test Set

- S&T developing 10+ artificial pedigrees (.cmf file format)
  - Comprised of biological relationship/kinship claims commonly encountered by USCIS and CBP (parent/child, grandparent/grandchild, siblings, etc)
  - Incorporates the 20 common paternity index formulas identified by AABB
  - Includes a wide number of alleles including off-ladder alleles and microvariants
  - Includes DNA profile complexities (e.g. parent-wrong gender, mutations, null alleles, and rare alleles)
- Enable quick and simple validation of Rapid DNA kinship software:
  - Verify allele frequencies & calculations
  - Verify correct reporting - inclusion/exclusion and probability of relationship
- Run every time expert software is updated/modified

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Technology is Field Ready!

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